

Echocardiographic Findings in 30 Children with Various Lysosomal Storage Disease

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BACKGROUND. Lysosomal storage diseases (LSD) can cause cardiovascular involvement, especially thickening of cardiac valves, ventricular hypertrophy and pulmonary hypertension. Little is known about differences of cardiac involvement in children between various LSD including Fabry disease (FD), mucopolysaccharidosis (MPS), Pompe disease (PD), I-cell disease (ICD) and mucopolipidosis (ML).

METHODS. The echocardiographic database was searched for all children with the diagnosis of LSD; all echocardiographic data were analyzed as well as clinical findings and information on enzyme replacement therapy (ERT) and bone marrow transplantation (BMT).

RESULTS. LSD in children is rare with only 30 patients (19 females) in our echocardiography data base. There were 21 pt with MPS (6 pt with type 1 MPS; 4 pt with type 2 MPS, 3 with type III MPS, 5 pt with type IV MPS, and 3 pt with Type VI MPS), 6 pt with FD, 2 pt with mucopolipidosis III, 1 pt with ICD, and 2 pt with PD. ERT was given in 5 pt with MPS and 3 pt with FD. Three pt with MPS had BMT.

The results of the echocardiographic examinations are shown in the Table. Any valvular heart disease was present in 18 pt. Mitral valve thickening was present in 18 pt, aortic valve thickening in 16 pt. In children valvular heart disease is significantly less often observed in FD than in MPS. Stenosis of the aortic valve was observed in 3 pt, and of the mitral valve in 2 pt (all with MPS). Cardiac symptoms were reported only in 2 pt with MPS. Valvular abnormalities were more common than left ventricular hypertrophy (2 pt).

	No of pt	Age, years	EF, %	LVMMI, g/m ²	LVH	Abnormal mitral valve	Abnormal aortic valve
MPS	21	11.2 ± 5.2*	59 ± 4	65 ± 39	1 (5%)	15 (71%)	14 (67%)
Fabry disease	6	8.4 ± 4.3	60 ± 4	67 ± 18	0	0	0
Other	5	4.9 ± 5.3	59 ± 3	48 ± 31*	1(20%)	3 (60%)	2 (40%)

No. = number; LVMMI = left ventricular muscle mass index; EF = ejection fraction; LVH = left ventricular hypertrophy; P < 0.05 is shown as *

CONCLUSION. Cardiovascular involvement in LSD in children is especially relevant in MPS. In FD in children, cardiac anomalies are rare. Especially in MPS, routine echocardiographic evaluation is necessary in children.

The figure (below) shows a parasternal long-axis view image of a 14 year boy with MPS type II (Hunter) with typical thickening of the mitral valve (arrow); LA = left atrium; LV = left ventricle

